Bill No. 7282 Written Testimony

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Hello,

My name is Maria Kaloidis. I am the Advocacy Chair for the Connecticut Chapter of Cure SMA. I would like to share with you an unforgettable moment two years ago when I came across a photo that changed my life. It was an image of a child standing tall, and dancing, extraordinarily similar to any other child. The caption was most memorable, it described that she was a girl with a disease called Spinal Muscular Atrophy Type I, and that she was able to receive treatment early in life. I can explain from personal experience that the outcome of a person with SMA without treatment and early intervention is much different than the image that I described. I am here today to advocate in favor of the proposed addition to Bill No. 7282, which would require every newborn infant in Connecticut be administered a screening test for Spinal Muscular Atrophy.

Spinal Muscular Atrophy or SMA is a genetic neurodegenerative disease which destroys the nerve cells in the spine responsible for voluntary movement, the motor neurons. Like other motor neuron disease such as ALS, the result is progressive weakness and muscle wasting. SMA robs individuals of the ability to walk, move, swallow, eat, speak, and breathe. In its most severe form SMA Type I, it is also the number one genetic cause of infant death. 1 in 11,000 babies will be born with SMA and 1 in 50 Americans is a carrier of the disease, so SMA is by no means uncommon. It is highly prevalent and a very real threat for newborn infants diagnosed with the disease.

As I said, my experience with the SMA is quite personal. I myself have SMA. I began showing symptoms of the disease a few weeks after birth and was not diagnosed until 9 months of age. The road to diagnosis was long, consisting of numerous hospital stays and misdiagnoses.

My parents had concerns very early on, beginning within the first month of my life and persisted to question the doctors when I failed to meet major motor milestones, such as rolling and crawling. I was described as being a lazy baby, which is a common experience among SMA families. After 9 months of searching, I was finally diagnosed with Spinal Muscular Atrophy Type I, which is the most severe form of the disease. The months of searching could have been easily avoided by a simple blood test which is used to detect the disease.

At the time of my diagnosis, I was given a prognosis of 2 years and only 5% chance of living past that age. Since that time, I have required increased support to live my life. I am currently 22 years old. At this stage in the progression of my illness, I consume most of my meals using a gastrointestinal tube because of my difficulty with swallowing and have about 25% of the lung capacity of the average patient. I have been on life support multiple times battling ventilator dependency because my decreased respiratory reserve makes me highly susceptible to life-threatening cases of pneumonia. I am very fortunate, however, to be on the strong end of the Type I spectrum. There is an even more serious reality for those on the other end of the spectrum. Most children with SMA type I will not live to see their second birthday, and should they live they will most likely be unable to speak, unable to sit upright, and dependent on machines, such as ventilators and G-tubes, to carry out their most basic life functions. Newborn screenings have the propensity to change the entire future of SMA because it will allow an individual to receive treatment in infancy. In SMA Type I, 80% of motor neuron death occurs in the first year of life.

The FDA approval of the first-ever treatment for SMA, called Spinraza, has completely revolutionized the expectations for those living with the disease and, when given early enough can produce the results in the image that I described in my introduction. Just like that girl, we

both share the same disease but have experienced vastly different outcomes. Individuals with SMA cannot reap the benefits of extraordinary therapies, like Spinraza, until a system of early detection of the disease is established. Over the past year, I began receiving treatment for my disease and have gained strength and made great strides, such as smiling with my whole face and speaking without getting winded, but am still quite affected by the disease because there were not any treatment available that I could receive in infancy. A treatment now does exist, we just need appropriate testing in Connecticut. Should this happen, SMA will no longer take lives in Connecticut. I hope that we can do better for the future of SMA. I hope you are compelled by those walking and dancing with SMA, like the girl dancing in that photo treated with Spinraza, and also by all of those with SMA type I who are still fighting the disease or were unable to receive treatment in time. SMA became a part of the Recommended Uniform Screening Panel on February 8, 2018, and I hope that we can see a similar result in Connecticut very soon. I ask you to please pass this proposal to include Spinal Muscular Atrophy on the newborn screening panel.

Thank you so much for your time and consideration!